Accredited Continuing Education Program Information

## Interpreting genetic tests: The basics of molecular diagnosis through application of results

## Course Organizers

**Ramiro Maldonado, MD**, Duke University
**Robert Hufnagel, MD, PhD**, National Institutes of Health (NIH)
**Kristy Lee, MS, CGC**, University of North Carolina (UNC)

## Course Description

Interpreting genetic testing results is a complex skill that requires an understanding of basic genetic principles, molecular techniques in genetic testing, and clinical implications of genes, variants, and phenotypes. Rapid advancements in genetics and genomics have created expertise gaps among healthcare providers and researchers for interpreting genetic testing for clinical purpose- however, understanding the distinct types of genetic testing and how to discern between pathogenic and benign variation variants is key in disease diagnosis, treatment, and prevention.

Attend this course if you are struggling with the interpretation of genetic testing results. Learn from experts in the field who will cover genetic testing and genetic counseling issues, as well as clinical information about syndromic and non-syndromic inherited ocular diseases Attendees will walk away with practical tips for the genetic diagnosis of inherited ocular diseases, and a comprehension of aspects ranging from ophthalmological evaluation to variant curation, including genetic counseling, research consortia, and public databases.

## Physician Accreditation Statement

The Association for Research in Vision and Ophthalmology (ARVO) is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians.

## Physician Credit Designation

ARVO designates this live activity for 3.75 *AMA PRA Category 1 Credits™.* Physicians should claim only the credit commensurate with the extent of their participation in the activity.

International Attendees: The American Medical Association (AMA) has determined that physicians not licensed in the U.S. who participate in CME activities are eligible for *AMA PRA Category 1 Credits™.*

## Statement of Need

Interpreting genetic testing results is a complex skill that requires an understanding of basic genetic principles, molecular techniques in genetic testing, and clinical implications of genes, variants, and phenotypes. Even after taking educational courses, clinicians of all specialties struggle with interpreting genetic testing results (Emily Edelman, National Society of Genetic Counselors 2019 annual meeting). Understanding the different types of genetic testing and how to discern between pathogenic and benign variation variant is key in disease diagnosis, treatment and prevention. Furthermore, gene therapies elevate target genes to “actionable” by the American College of Medical Genetics and Genomics (ACMGG) and the Clinical Genome Resource (ClinGen).

## Target Audience

Ophthalmologists, Genetic counselors, Geneticists, Researchers and Trainees (residents, fellows, postdocs) interested in genetic testing and gene therapies related to ophthalmic diseases and disorders.

## Educational Objectives

After participating in this CME activity, participants should be able to:

* Describe the molecular basis of genetic testing
* Classify the most common syndromic and non-syndromic inherited ocular diseases
* Interpret positive, inconclusive and negative genetic testing results
* Apply new concepts to utilize public databases for gene and variant curation

## European Union of Medical Specialists (UEMS) CME/CPD Requirements

European physicians can convert CME credit from the course to meet European Union of Medical Specialists (UEMS) CME/CPD requirements.

The American Medical Association has an agreement of mutual recognition of CME credits with the UEMS, the accreditation body for European countries. Physicians interested in converting *AMA PRA Category 1 Credit™* to UEMS-European Accreditation Council for Continuing Medical Education CME credits (ECMECs) should contact the UEMS at mutualrecognition@uems.eu.

## CME Credits and Certificates Fee

Learners who register for “CME Credits and Certificates” as part of their Annual Meeting registration are eligible to claim *AMA PRA Category 1 Credit™* for participating in this course. This fee is paid once and covers credit claiming for an Education Course and the Annual Meeting.

|  |  |  |
| --- | --- | --- |
| **CME credits and certificate fee** | **By March 15** | **After March 15** |
| ARVO MIT/Nonmember-in-Training | $ 79 | $ 79 |
| ARVO Member/Exhibitor (paid full-meeting access badge)  | $ 99 | $ 119 |
| Nonmember/Exhibitor (complimentary full meeting access badge only) | $ 99 | $ 119 |

## Instructions to receive credit and/or certificates

Learners interested in claiming *AMA PRA Category 1 Credits™* or obtain a Certificate of Participation for participating in the course must pay the CME credits and Certificates fee as part of their Annual Meeting Registration. Payment is collected during the Annual Meeting registration process by adding “CME credits and Certificates” as a session. Attendees who do not register for CME before the conclusion of the meeting will not be able to claim and request credit. Learners registered as guests are not eligible to receive CME credit.

Credits and certificates are awarded within our ARVOLearn platform. CME registrants will receive an email with instructions prior to the start of the course or within two business days of your purchase (whichever is later). All requests for credit must be submitted no later than 11:59pm U.S. Eastern Time on Friday, Aug. 30, 2023.

## Certificate of Participation

ARVO is accredited to offer *AMA PRA Category 1 Credits™* to physicians. All other participants in the course may receive a Certificate of Participation that documents the number of CME hours attended\* by following the same instructions listed above and paying the CME credits and certificates fee.

\*Some accrediting agencies may award equivalent Continuing Education (CE) credit when presented with a Certificate of Participation from an activity that offered *AMA PRA Category 1 Credit™.* Check with your accrediting agency for information.

## Certificate of Attendance

Certificates of attendance are available free to all attendees. These certificates state that one has attended the course; but does not offer credit tracking. They will be available for pick-up onsite at the meeting registration desk and ARVO Central in the exhibit hall.

## Contact Us

ARVO
5515 Security Lane, Suite 500
Rockville, Md. 20852, USA
Email: education@arvo.org, p. +1.240.221.2900

## Disclaimer

Education Courses provide a forum for the open exchange and discussion of research results and scientific advancements in the field of ophthalmology; however, ARVO makes no representation or warranty as to the truth, originality, or accuracy of the information presented at courses or in materials distributed in connection with them. Nor are the views expressed by the individual speakers necessarily the views of ARVO. ARVO supports the ACCME’s policy on evidence-based content and encourages faculty to adhere to these standards when planning a presentation.

Participants have an implied responsibility to use the newly acquired information to enhance patient outcomes and their own professional development. The information presented in this course is not meant to serve as a guideline for patient management. Any procedures, medications, or other courses of diagnosis or treatment discussed in this activity should not be used by clinicians without evaluation of patient conditions and possible contraindications on dangers in use, review of any applicable manufacturer’s product information, and comparison with recommendations of other authorities.

## Disclaimer off-label use

Education Courses may contain discussion of published and/or investigational uses of agents that are not indicated by the FDA. ARVO does not recommend the use of any agent outside of the labeled indications.

The opinions expressed in the educational activity are those of the faculty and do not necessarily represent the views of any organization associated with this activity. Please refer to the official prescribing information for each product for discussion of approved indications, contraindications and warnings.

## Disclosure of Relevant Financial Relationships

ARVO requires instructors, planners, managers and other individuals who are in a position to control the content of this activity to disclose all financial relationships with any ineligible company within the last 24 months. All identified relevant financial relationships are thoroughly vetted by ARVO for fair balance, scientific objectivity of studies mentioned in the materials or used as the basis for content, and appropriateness of patient care recommendations. All relevant financial relationships have been mitigated by ARVO.

ARVO considers presenting authors, not co-authors, to be in control of the educational content. It is ARVO’s policy and traditional scientific publishing practice to acknowledge all people contributing to the research, regardless of CME control of the live presentation of that content. ARVO may collect and publish disclosures from co-authors although they do not have control of the CME content.

**Disclosures for all individuals involved in planning and managing this accredited CE activity are listed here with their relevant disclosure information. Disclosure information for presenters and course moderators will be provided within a disclosure slide(s), prior to the start of the course and their respective presentations.**

*The following individuals on the Professional Development and Education Committee (PDEC) reported no relevant financial relationships.*Alison Abraham, PhD; Maria Fernanda Abalem, MD, MSc; Radwan Ajlan, MBBCh, FRCSC; Lin Cheng, MD, PhD; Ngozi Chidi-Egboka, OD, MPH, FNCO, FAAO; Helene Filipe, MD; Renu Kowluru, PhD, FARVO; Emily Patterson, PhD; Daisy Shu, PhD; Menglu Yang, MD, PhD

*The following individuals on the Continuing Medical Education Committee (CME) reported no relevant financial relationships.*Parisa Emami, MD, MPH; Shilpa Kodati, MBBS; Wendy Liu, MD, PhD; Yannis Paulus, MD; Sangly Srinivas, PhD; Mary Whitman, MD, PhD

*The following course organizers and activity managers reported no relevant financial relationships.*Robert Hufnagel, MD, PhD (course organizer), Kristy Lee, MS, CGC, University of North Carolina (UNC) (course organizer), Cathy Conley, (activity manager)

*The following clinical content reviewers reported no relevant financial relationships.*Lindsay Scott, PT, DPT, ATC

*The following activity planners and managers reported relevant financial relationships.*

|  |  |  |
| --- | --- | --- |
| **Full name** | **Role** | **Disclosure**  |
| Ramiro Maldonado, MD | Course Organizer | ProQR Therapeutics: (C) |
| Priya Chaudhary, PhD | PDEC | Alcon Research Institue (F) |
| Azza El-Remessy, PhD, PharmD, BC-ADM | PDEC | Nour Therapeutics, LLC: (O) |
| William Foster, MD, PhD, FRCSC | PDEC | Altasciences (C); Lynthera (C); US Patent 9,050,171-Small diameter fragmatome for minimally traumatic retained lens fragments removal (P) |
| Edmund Tsui, MD | PDEC | Kowa Company Ltd (C, F); Cylite Pty Ltd (F); EyePoint Pharmaceuticals (C) |
| Thomas Ach, MD, FEBO | CME | Apellis Pharmaceutical: (R); Roche: (R); Novartis: (R, F); Bayer: (R); Heidelberg Engineering: (R); MacRegen Inc.: (I) |
| Ron Adelman, MD, MPH, MBA | CME | Tesseract: (C); Johnson & Johnson/Jenssen: (C) |
| Natasha Kolomeyer, MD | CME | AbbVie/Allergan: (F, C); Guardion Health Services Inc.: (F); Equinox: (F); Nicox: (F); Olleyes: (F); Santen: (F); Glaukos: (F); Diopsys: (F); Aerie: (F); |
| Louis Pasquale, MD | CME | Eyenovia: (C); Twenty Twenty: (C); Skye Biosciences: (C) |
| Laura Lanford | Staff | Johnson and Johnson: (I); Pfizer: (I) |

*Financial relationship disclosure code definitions*

**C (Consultant/Contractor)** Indicates you are a consultant or independent contractor (including contracted research) for an ACCME-defined ineligible company.

**E (Employment)** Indicates you are an employee of an ACCME-defined ineligible company.

**F (Financial Support)** Indicates financial support received from an ACCME-defined ineligible company, in the form of research funding (even if the institution receives the research grant and manages the funds), grants, research materials or in-kind services (e.g., sample analysis).

**I (Personal Financial Interest)** Indicates you are an investor in a company or competing company, including an ACCME-defined ineligible company, other than through a mutual or retirement fund.

**N (No Financial Relationship)** Indicates there is no financial relationship to disclose.

**O (Owner/Co-Owner/Founder/Co-Founder)** Indicates you are an owner, co-owner, founder and/or co-founder of an ACCME-defined ineligible company.

**P (Patent)** Indicates you are an inventor/developer designated on a patent, patent application, copyright, or trade secret, whether the patent, copyright, etc. is presently licensed or otherwise commercialized, or could be in competition with the technology described.

**R (Recipient)** Indicates you have received gifts, honoraria, travel reimbursement, patent royalties, or any other financial compensation in any amount from an ACCME-defined ineligible company.

**S (non-remunerative)** Indicates you have position(s) of influence such as officer, board member, trustee, spokesperson, etc.